

Adrenoleukodystrophy: What You Need to Know

Adrenoleukodystrophy is a disorder that affects the nervous system, adrenal glands and testes. In this handout, you will learn about the disease and what causes it. You will also learn how doctors test for and treat adrenoleukodystrophy.

WHAT IS ADRENOLEUKODYSTROPHY?

Adrenoleukodystrophy is a genetic disorder that affects the nervous system, **adrenal glands** (small glands on top of the kidneys) and **testes**. It causes chemicals called **very long chain fatty acids** to build up in the body. These fatty acids damage the adrenal glands and **myelin sheaths** (the protective covering of nerves).

WHO CAN GET ADRENOLEUKODYSTROPHY?

Adrenoleukodystrophy mostly affects males. In rare cases, it can also affect females. Symptoms can show anytime between childhood and age 50.

WHAT CAUSES ADRENOLEUKODYSTROPHY?

Adrenoleukodystrophy is caused by a mutation (change) in the *ABCD1* gene.

WHAT ARE SYMPTOMS OF ADRENOLEUKODYSTROPHY?

Symptoms depend on how far along the disease has progressed (which parts of the body are affected):

- **In the mildest form**, people have adrenal damage in the childhood or teenage years. The nerves might become damaged later in life.
- **In the intermediate form**, nerve, brain and behavioral symptoms start in early adulthood and middle age. Many have adrenal damage.
- **In the most severe (cerebral) form**, children develop learning and behavioral problems between 4-10 years of age. Over time, they may have difficulty reading, writing and understanding speech and written material. They might also have adrenal gland damage.

HOW DO DOCTORS TEST FOR ADRENOLEUKODYSTROPHY?

Doctors use genetic and hormone testing and brain scans to test for adrenoleukodystrophy. Genetic testing is usually done through a blood test.

HOW DO DOCTORS TREAT ADRENOLEUKODYSTROPHY?

Treatment depends on a person's symptoms and the type of adrenoleukodystrophy that they have. It is important to diagnose the disorder early so treatment will work better. Your child's doctor will help figure out the best treatment.

- **Steroid replacement therapy**
This is necessary when the adrenal glands are damaged and can no longer make adrenal hormones cortisol and aldosterone.
- **Lorenzo's Oil**
Along with reduced fat intake, Lorenzo's oil has been used to reduce the risk of progression of the severe cerebral form of the disease
- **Stem cell transplant**
Stem cells are cells from bone marrow (soft core of bones) that can help make more protein from the *ABCD1* gene.

WHAT IS THE OUTCOME FOR A CHILD WITH ADRENOLEUKODYSTROPHY?

The outcome depends on which form of the disease your child has. We understand this is a difficult time for you. Your child's care team is here with you every step of the way. We can answer any questions and address your concerns at any time.